

Illinois Department of Public Health
Newborn Screening Laboratory Subcommittee
Illinois Department of Public Health, Division of Laboratories
2121 W. Taylor St., Chicago, Illinois
Teleconference
Meeting Minutes: January 25, 2017

Subcommittee Members Attending:

George Hoganson-University of Illinois at Chicago – Chair
Barbara Burton- Lurie Children’s Hospital
Denise Lonigro-Advocate Christ Hospital
Gopal Srinivasan-Mt. Sinai Hospital
Ramsay Fuleihan- Lurie Children’s Hospital
Kristin Clemenz-Lurie Children’ Hospital

IDPH Staff:

Matt Charles, Rong Shao, Khaja Basheeruddin, Rich Zimmerman, Arthur Kohrman, Molly Lamb, Claudia Nash, Heather Shryock

Welcome

The meeting was called to order at 9:05 AM, followed by a review of committee members and introductions. Dr. Hoganson, Subcommittee Chair, indicated that the purpose of this committee is to support IDPH staff. Since the committee has not met recently, and with recent laboratory staffing changes, he stressed it is probably a good time to review data, how cases are reported and to discuss cutoffs and test interpretation as well as any existing technical issues. Dr. Hoganson also indicated that there has been an increase in the number of positive cases reported, and Dr. Burton stated there has been confusion regarding some cases with analytes in the normal range that are being reported as abnormal.

Overview of Laboratory and Follow-up Issues

Matt Charles indicated that he is working to fill the vacant position of Newborn Screening Division Chief. The Newborn Screening Laboratory has also lost some supervisory positions due to retirement, but they have reassigned staff to handle those duties. Claudia Nash indicated that there are some vacancies in the follow-up program as well, including a nurse supervisor. Follow up staff also have additional demands due to participation in more monthly teleconference meetings sponsored by the NewSTEPs newborn screening technical assistance program of the Association of Public Health Laboratories, and additional requests for data.

Review of Data

Dr. Hoganson indicated the revised protocol related to timing of specimen collection for premature and sick newborns has created some confusion with pediatricians, and an increase in positive test results. The recommendation by the Clinical and Laboratory Standards Institute (CLSI) to have a specimen collected on every newborn upon admission to the NICU or special care nursery is now being recommended in Illinois to assure that a baseline sample is collected prior to the initiation of blood transfusion or special feeding. Dr. Hoganson stated the laboratory should only be reporting test results that are meaningful in the first 24 hours of life, rather than reporting any abnormal finding, since it is known that certain analytes will be abnormal due to early specimen collection. This issue will be reviewed further during the next subcommittee meeting.

Screening data for the period of 2013-2016 was reviewed. While stability is noted during this time for the screening data for most disorders (biotinidase deficiency, CAH, sickle cell, and CF), the birth rate and diagnosed cases, the number of positive newborn screens increased significantly for galactosemia, amino, organic and fatty acid oxidation disorders. The NBS laboratory implemented a new testing platform (GSP-genetic screening processor) which tests for total galactose and GALT, and which is more user friendly and efficient and allows for multiple assays (galactosemia, CF, biotinidase deficiency, hypothyroidism and CAH) to be conducted on the same instrument. The data reviewed indicate a large increase in the specimens with total GAL <14 mg/dL and reduced GALT (≤ 3.4 U/dL). The current recommendation for these cases is referral to a specialist, but frequently physicians decide to simply repeat the newborn screen and close the case if normal. Dr. Hoganson indicated that carriers might be missed, but questioned if the intent of the screen is to identify carriers. Dr. Hoganson asked for this data to be analyzed further prior to the next subcommittee meeting to differentiate specimens with varying levels in this category. Dr. Hoganson also asked for the rationale for the total GAL cutoff of 14 mg/dL, and he indicated that some infants diagnosed with galactosemia have levels less than 14. The lab will present more data at the next subcommittee meeting.

With regard to the amino, organic and fatty acid data for 2013-2016, the number of specimens with positive screens for certain analytes have increased significantly over time. It was suggested by Dr. Hoganson to examine the data further for premature newborns, and specimens collected prior to 24 hours of life. He also stated that age related cutoffs are needed, and that strategies need to be implemented to limit false positive.

Testing for SCID is going well with 13 cases identified since screening began in June 2014, and other T cell lymphopenias also identified. Dr. Fuleihan stated that the immunologists did not expect to identify so many idiopathic T cell lymphopenias through screening, and that we could consider changing the cutoff slightly, but might then miss some DiGeorge cases. Illinois uses the same screening method as Massachusetts and has the same cutoff of 250 TRECs.

Lysosomal Storage Disorders- Review of Pilot Test Implementation

The LSD pilot began November 3, 2014, with newborns from four hospitals being tested for five LSDs; Pompe, Gaucher, Fabry, MPS I and Niemann-Pick. Additional hospitals have been added to the pilot, and currently all specimens from 8 hospitals, including Prentice Women's Hospital, are included. To date, approximately 4,700 samples have been tested with 14 newborns having an abnormal finding on screening, and no cases diagnosed.

Regarding Krabbe molecular testing, attempts to secure a contract with the New York state laboratory have failed, due to inconsistencies between IDPH and NY attorneys regarding the required contract language. A contractual arrangement is being finalized through the University of Illinois at Chicago to send samples to Mayo laboratory for this testing. Dr. Dizikes stated that Mayo has agreed to perform psychosine and 30kb deletion testing on all samples submitted, with a 24 hour turn-around time, and will conduct sequencing for specimens as needed, with a proposed 7 day turn-around time. Dr. Hoganson stated, that since this deviates from the protocol originally approved by the LSD subcommittee, this change should be reviewed and approved at the next meeting of the LSD subcommittee, as well as the timing for how and when results should be reported by IDPH. It is anticipated that Krabbe will be added to the pilot prior to further expansion of testing specimens for LSDs from other hospitals.

Review of 2013/2014 Data

Abnormal screening data and diagnosed case data were compared for 2013 and 2014. During 2014, there were approximately 100 more samples with a positive amino acid screening result and 155 more with an abnormal fatty acid oxidation result than in 2013, although there were 32% fewer diagnosed cases in these categories in 2014. Other categories were comparable for the two years.

Some pediatricians and specialists have questioned why IDPH is reporting test results as “presumptive positive” for some amino, fatty acid and organic acid analytes that are in the normal range (all of these cases have been normal with further diagnostic testing). Dr. Dizikes indicated other ratios are abnormal in these cases, but no explanation has been provided on the report to physicians. Dr. Dizikes also indicated there are other cutoffs used for older babies. Dr. Hoganson requested that an explanatory note be added to these reports to lessen the confusion for physicians. Dr. Hoganson and Dr. Kohrman also requested that IDPH compare screening/diagnosed case data with that in other states and to the Illinois birth rate.

Information regarding newborn screening testing turn-around time was reviewed for December 2014 to February 2015, which indicates a mean time of 1.6 days from specimen collection to lab receipt, 4.1 days from receipt to report of abnormal results and 9.4 days from specimen receipt to report of normal results.

Adrenoleukodystrophy

Claudia Nash indicated that legislation has been introduced to add X-linked adrenoleukodystrophy (ALD) to the Illinois newborn screening panel. IDPH staff have gathered information from New York, the only state screening for ALD, and the IDPH Director, Dr. Shah, has been discussing this issue with the legislative sponsor. Further information about ALD newborn screening was provided at the February 2015 meeting of the HHS Discretionary Advisory Committee for Heritable Disorders in Newborns and Children, which may vote on the inclusion of ALD later this summer.

Timeliness Issues

Recommendations were made at the February 2015 meeting of the HHS Discretionary Advisory Committee for Heritable Disorders in Newborns and Children, regarding five areas related to newborn screening timeliness. State newborn screening programs are being encouraged to address these areas, with a goal of 95% compliance by 2017. They are:

- 1- All initial specimens should be collected by 48 hours of life
- 2- All specimens should be received in lab within 24 hours of collection
- 3- Results of all time critical tests should be reported by the 5th day of life
- 4- Results of all presumptive positive results should be reported by the 7th day of life
- 5- All tests should be completed by the 7th day of life

Newborn screening staff will track these metrics on a quarterly basis beginning with a baseline of the fourth quarter of calendar year 2014.

IT System Developments

Birth Related Data System

Progress is being made toward developing an interface between the newborn screening data system and the vital records (birth certificate) data system. This will allow for direct population of information from the birth record into the newborn screening system, which will yield more accurate and more complete information, as well as document that a newborn screening specimen has been received for each birth. Testing is planned for the summer of 2015 at the two birth hospitals in Springfield.

Direct Access to Newborn Screening Results by Provider (eReports)

A feature in the newborn screening data system is being developed which would allow hospitals and physicians to look up and print newborn screening results for their patients. It is anticipated this feature may be available by late summer 2015.

The meeting adjourned at 10:05 a.m.